Katherine L Nathanson

List of Publications by Year
in descending order

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347 papers	34,049 citations	3525 90 h-index	4545 171 g-index
363 all docs	363 docs citations	363 times ranked	42371 citing authors

#	Article	IF	Citations
1	Clinical efficacy of a RAF inhibitor needs broad target blockade in BRAF-mutant melanoma. Nature, 2010, 467, 596-599.	13.7	1,610
2	The Tuberous Sclerosis Complex. New England Journal of Medicine, 2006, 355, 1345-1356.	13.9	1,570
3	T-cell invigoration to tumour burden ratio associated with anti-PD-1 response. Nature, 2017, 545, 60-65.	13.7	1,280
4	Acquired Resistance to BRAF Inhibitors Mediated by a RAF Kinase Switch in Melanoma Can Be Overcome by Cotargeting MEK and IGF-1R/PI3K. Cancer Cell, 2010, 18, 683-695.	7.7	1,139
5	Low-penetrance susceptibility to breast cancer due to CHEK2*1100delC in noncarriers of BRCA1 or BRCA2 mutations. Nature Genetics, 2002, 31, 55-59.	9.4	1,001
6	Rare cell variability and drug-induced reprogramming as a mode of cancer drug resistance. Nature, 2017, 546, 431-435.	13.7	938
7	Gene-Panel Sequencing and the Prediction of Breast-Cancer Risk. New England Journal of Medicine, 2015, 372, 2243-2257.	13.9	764
8	Cancer Risk Estimates for BRCA1 Mutation Carriers Identified in a Risk Evaluation Program. Journal of the National Cancer Institute, 2002, 94, 1365-1372.	3.0	611
9	Network modeling links breast cancer susceptibility and centrosome dysfunction. Nature Genetics, 2007, 39, 1338-1349.	9.4	602
10	Comprehensive Molecular Characterization of Pheochromocytoma and Paraganglioma. Cancer Cell, 2017, 31, 181-193.	7.7	532
11	Multiple independent variants at the TERT locus are associated with telomere length and risks of breast and ovarian cancer. Nature Genetics, 2013, 45, 371-384.	9.4	493
12	PTEN Loss Confers BRAF Inhibitor Resistance to Melanoma Cells through the Suppression of BIM Expression. Cancer Research, 2011, 71, 2750-2760.	0.4	488
13	Metastatic potential of melanomas defined by specific gene expression profiles with no BRAF signature. Pigment Cell & Melanoma Research, 2006, 19, 290-302.	4.0	483
14	A single dose of neoadjuvant PD-1 blockade predicts clinical outcomes in resectable melanoma. Nature Medicine, 2019, 25, 454-461.	15.2	466
15	HIF-Î± Effects on c-Myc Distinguish Two Subtypes of Sporadic VHL-Deficient Clear Cell Renal Carcinoma. Cancer Cell, 2008, 14, 435-446.	7.7	441
16	A Population-Based Study of Genes Previously Implicated in Breast Cancer. New England Journal of Medicine, 2021, 384, 440-451.	13.9	414
17	Association of Type and Location ofBRCA1andBRCA2Mutations With Risk of Breast and Ovarian Cancer. JAMA - Journal of the American Medical Association, 2015, 313, 1347.	3.8	390
18	A Functional Genomic Approach Identifies FAL1 as an Oncogenic Long Noncoding RNA that Associates with BMI1 and Represses p21 Expression in Cancer. Cancer Cell, 2014, 26, 344-357.	7.7	361

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19	Identification of 12 new susceptibility loci for different histotypes of epithelial ovarian cancer. Nature Genetics, 2017, 49, 680-691.	9.4	356
20	Integrated Molecular Characterization of Testicular Germ Cell Tumors. Cell Reports, 2018, 23, 3392-3406.	2.9	324
21	Common variation in KITLG and at 5q31.3 predisposes to testicular germ cell cancer. Nature Genetics, 2009, 41, 811-815.	9.4	319
22	A locus on 19p13 modifies risk of breast cancer in BRCA1 mutation carriers and is associated with hormone receptorâ€“negative breast cancer in the general population. Nature Genetics, 2010, 42, 885-892.	9.4	309
23	Phase I trial of hydroxychloroquine with dose-intense temozolomide in patients with advanced solid tumors and melanoma. Autophagy, 2014, 10, 1369-1379.	4.3	309
24	Opposing Functions of Interferon Coordinate Adaptive and Innate Immune Responses to Cancer Immune Checkpoint Blockade. Cell, 2019, 178, 933-948.e14.	13.5	301
25	Two Decades After BRCA: Setting Paradigms in Personalized Cancer Care and Prevention. Science, 2014, 343, 1466-1470.	6.0	300
26	Identification of ten variants associated with risk of estrogen-receptor-negative breast cancer. Nature Genetics, 2017, 49, 1767-1778.	9.4	289
27	Increased cyclin D1 expression can mediate BRAF inhibitor resistance in BRAF V600Eâ€“mutated melanomas. Molecular Cancer Therapeutics, 2008, 7, 2876-2883.	1.9	284
28	Molecular Stratification of Clear Cell Renal Cell Carcinoma by Consensus Clustering Reveals Distinct Subtypes and Survival Patterns. Genes and Cancer, 2010, 1, 152-163.	0.6	283
29	A recurrent germline PAX5 mutation confers susceptibility to pre-B cell acute lymphoblastic leukemia. Nature Genetics, 2013, 45, 1226-1231.	9.4	270
30	Cancer Risks Associated With Germline PALB2 Pathogenic Variants: An International Study of 524 Families. Journal of Clinical Oncology, 2020, 38, 674-685.	0.8	270
31	ENIGMA-Evidence-based network for the interpretation of germline mutant alleles: An international initiative to evaluate risk and clinical significance associated with sequence variation in BRCA1 and BRCA2 genes. Human Mutation, 2012, 33, 2-7.	1.1	269
32	Genome-wide association study identifies 32 novel breast cancer susceptibility loci from overall and subtype-specific analyses. Nature Genetics, 2020, 52, 572-581.	9.4	265
33	Counselling framework for moderate-penetrance cancer-susceptibility mutations. Nature Reviews Clinical Oncology, 2016, 13, 581-588.	12.5	258
34	Common Breast Cancer-Predisposition Alleles Are Associated with Breast Cancer Risk in BRCA1 and BRCA2 Mutation Carriers. American Journal of Human Genetics, 2008, 82, 937-948.	2.6	257
35	Genome-Wide Association Study in BRCA1 Mutation Carriers Identifies Novel Loci Associated with Breast and Ovarian Cancer Risk. PLoS Genetics, 2013, 9, e1003212.	1.5	244
36	A meta-analysis identifies new loci associated with body mass index in individuals of African ancestry. Nature Genetics, 2013, 45, 690-696.	9.4	232

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37	Pheochromocytoma: The Expanding Genetic Differential Diagnosis. Journal of the National Cancer Institute, 2003, 95, 1196-1204.	3.0	230
38	Mutational spectrum in a worldwide study of 29,700 families withBRCA1orBRCA2mutations. Human Mutation, 2018, 39, 593-620.	1.1	224
39	Identification of six new susceptibility loci for invasive epithelial ovarian cancer. Nature Genetics, 2015, 47, 164-171.	9.4	221
40	RAD51 135Gâ†'C Modifies Breast Cancer Risk among BRCA2 Mutation Carriers: Results from a Combined Analysis of 19 Studies. American Journal of Human Genetics, 2007, 81, 1186-1200.	2.6	217
41	The Mitogen-Activated Protein/Extracellular Signal-Regulated Kinase Kinase Inhibitor AZD6244 (ARRY-142886) Induces Growth Arrest in Melanoma Cells and Tumor Regression When Combined with Docetaxel. Clinical Cancer Research, 2008, 14, 230-239.	3.2	214
42	Measurements of Tumor Cell Autophagy Predict Invasiveness, Resistance to Chemotherapy, and Survival in Melanoma. Clinical Cancer Research, 2011, 17, 3478-3489.	3.2	213
43	BRCA locus-specific loss of heterozygosity in germline BRCA1 and BRCA2 carriers. Nature Communications, 2017, 8, 319.	5.8	212
44	Von Hippelâ€"Lindau and Hereditary Pheochromocytoma/Paraganglioma Syndromes: Clinical Features, Genetics, and Surveillance Recommendations in Childhood. Clinical Cancer Research, 2017, 23, e68-e75.	3.2	205
45	The Y Deletion gr/gr and Susceptibility to Testicular Germ Cell Tumor. American Journal of Human Genetics, 2005, 77, 1034-1043.	2.6	197
46	Uterine Cancer After Risk-Reducing Salpingo-oophorectomy Without Hysterectomy in Women With BRCA Mutations. JAMA Oncology, 2016, 2, 1434.	3.4	189
47	Inherited Mutations in Pheochromocytoma and Paraganglioma: Why All Patients Should Be Offered Genetic Testing. Annals of Surgical Oncology, 2013, 20, 1444-1450.	0.7	182
48	Pheochromocytoma and paraganglioma: understanding the complexities of the genetic background. Cancer Genetics, 2012, 205, 1-11.	0.2	177
49	HIF2Î± inhibition promotes p53 pathway activity, tumor cell death, and radiation responses. Proceedings of the National Academy of Sciences of the United States of America, 2009, 106, 14391-14396.	3.3	176
50	Molecular Profiling of Patient-Matched Brain and Extracranial Melanoma Metastases Implicates the PI3K Pathway as a Therapeutic Target. Clinical Cancer Research, 2014, 20, 5537-5546.	3.2	169
51	Tumor Genetic Analyses of Patients with Metastatic Melanoma Treated with the BRAF Inhibitor Dabrafenib (GSK2118436). Clinical Cancer Research, 2013, 19, 4868-4878.	3.2	167
52	Concurrent MEK2 Mutation and BRAF Amplification Confer Resistance to BRAF and MEK Inhibitors in Melanoma. Cell Reports, 2013, 4, 1090-1099.	2.9	162
53	Variants at 6q21 implicate PRDM1 in the etiology of therapy-induced second malignancies after Hodgkin's lymphoma. Nature Medicine, 2011, 17, 941-943.	15.2	155
54	Meta-analysis identifies four new loci associated with testicular germ cell tumor. Nature Genetics, 2013, 45, 680-685.	9.4	154

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55	Prediction of Breast and Prostate Cancer Risks in Male BRCA1 and BRCA2 Mutation Carriers Using Polygenic Risk Scores. Journal of Clinical Oncology, 2017, 35, 2240-2250.	0.8	152
56	Screening for Genomic Rearrangements in Families with Breast and Ovarian Cancer Identifies BRCA1 Mutations Previously Missed by Conformation-Sensitive Gel Electrophoresis or Sequencing. American Journal of Human Genetics, 2000, 67, 841-850.	2.6	149
57	Conflicting Interpretation of Genetic Variants and Cancer Risk by Commercial Laboratories as Assessed by the Prospective Registry of Multiplex Testing. Journal of Clinical Oncology, 2016, 34, 4071-4078.	0.8	147
58	Whole-exome sequencing identifies somatic ATRX mutations in pheochromocytomas and paragangliomas. Nature Communications, 2015, 6, 6140.	5.8	143
59	Platinum response characteristics of patients with pancreatic ductal adenocarcinoma and a germline BRCA1, BRCA2 or PALB2 mutation. British Journal of Cancer, 2020, 122, 333-339.	2.9	141
60	Genome-wide linkage screen for testicular germ cell tumour susceptibility loci. Human Molecular Genetics, 2006, 15, 443-451.	1.4	138
61	Biallelic TSC gene inactivation in tuberous sclerosis complex. Neurology, 2010, 74, 1716-1723.	1.5	134
62	Association of HPC2/ELAC2 Genotypes and Prostate Cancer. American Journal of Human Genetics, 2000, 67, 1014-1019.	2.6	133
63	Factors Determining Dissemination of Results and Uptake of Genetic Testing in Families with Known BRCA1/2 Mutations. Genetic Testing and Molecular Biomarkers, 2008, 12, 81-91.	1.7	130
64	Prevalence of mutations in a panel of breast cancer susceptibility genes in BRCA1/2-negative patients with early-onset breast cancer. Genetics in Medicine, 2015, 17, 630-638.	1.1	128
65	Care of adults with neurofibromatosis type 1: a clinical practice resource of the American College of Medical Genetics and Genomics (ACMG). Genetics in Medicine, 2018, 20, 671-682.	1.1	128
66	Breast cancer risk variants at 6q25 display different phenotype associations and regulate ESR1, RMND1 and CCDC170. Nature Genetics, 2016, 48, 374-386.	9.4	125
67	A second independent locus within DMRT1 is associated with testicular germ cell tumor susceptibility. Human Molecular Genetics, 2011, 20, 3109-3117.	1.4	124
68	Biallelic Deleterious BRCA1 Mutations in a Woman with Early-Onset Ovarian Cancer. Cancer Discovery, 2013, 3, 399-405.	7.7	124
69	Hereditary Kidney Cancer Syndromes. Advances in Chronic Kidney Disease, 2014, 21, 81-90.	0.6	122
70	Recommendations for Cancer Surveillance in Individuals with RASopathies and Other Rare Genetic Conditions with Increased Cancer Risk. Clinical Cancer Research, 2017, 23, e83-e90.	3.2	122
71	Fine-mapping of 150 breast cancer risk regions identifies 191 likely target genes. Nature Genetics, 2020, 52, 56-73.	9.4	120
72	Variants in CHEK2 Other than 1100delC Do Not Make a Major Contribution to Breast Cancer Susceptibility. American Journal of Human Genetics, 2003, 72, 1023-1028.	2.6	119

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73	A Comprehensive Patient-Derived Xenograft Collection Representing the Heterogeneity of Melanoma. Cell Reports, 2017, 21, 1953-1967.	2.9	117
74	Evaluation of ACMG-Guideline-Based Variant Classification of Cancer Susceptibility and Non-Cancer-Associated Genes in Families Affected by Breast Cancer. American Journal of Human Genetics, 2016, 98, 801-817.	2.6	113
75	Phase II Study of Maintenance Rucaparib in Patients With Platinum-Sensitive Advanced Pancreatic Cancer and a Pathogenic Germline or Somatic Variant in BRCA1, BRCA2, or PALB2. Journal of Clinical Oncology, 2021, 39, 2497-2505.	0.8	113
76	A genome wide linkage search for breast cancer susceptibility genes. Genes Chromosomes and Cancer, 2006, 45, 646-655.	1.5	111
77	Determination of Cancer Risk Associated with Germ Line BRCA1 Missense Variants by Functional Analysis. Cancer Research, 2007, 67, 1494-1501.	0.4	110
78	Personalized Preclinical Trials in BRAF Inhibitorâ€“Resistant Patient-Derived Xenograft Models Identify Second-Line Combination Therapies. Clinical Cancer Research, 2016, 22, 1592-1602.	3.2	108
79	The contribution of pathogenic variants in breast cancer susceptibility genes to familial breast cancer risk. Npj Breast Cancer, 2017, 3, 22.	2.3	108
80	Population Frequency of Germline BRCA1/2 Mutations. Journal of Clinical Oncology, 2016, 34, 4183-4185.	0.8	107
81	Identification of a BRCA2-Specific Modifier Locus at 6p24 Related to Breast Cancer Risk. PLoS Genetics, 2013, 9, e1003173.	1.5	105
82	Meta-analysis of five genome-wide association studies identifies multiple new loci associated with testicular germ cell tumor. Nature Genetics, 2017, 49, 1141-1147.	9.4	105
83	Phase II Trial of Temozolomide and Sorafenib in Advanced Melanoma Patients with or without Brain Metastases. Clinical Cancer Research, 2009, 15, 7711-7718.	3.2	104
84	Active Notch1 Confers a Transformed Phenotype to Primary Human Melanocytes. Cancer Research, 2009, 69, 5312-5320.	0.4	103
85	A Classification Model for BRCA2 DNA Binding Domain Missense Variants Based on Homology-Directed Repair Activity. Cancer Research, 2013, 73, 265-275.	0.4	103
86	PALB2 mutations in familial breast and pancreatic cancer. Familial Cancer, 2011, 10, 225-231.	0.9	102
87	Large scale multifactorial likelihood quantitative analysis of BRCA1 and BRCA2 variants: An ENIGMA resource to support clinical variant classification. Human Mutation, 2019, 40, 1557-1578.	1.1	102
88	The Relative Contribution of Point Mutations and Genomic Rearrangements in BRCA1 and BRCA2 in High-Risk Breast Cancer Families. Cancer Research, 2008, 68, 7006-7014.	0.4	100
89	Common variants in LSP1, 2q35 and 8q24 and breast cancer risk for BRCA1 and BRCA2 mutation carriers. Human Molecular Genetics, 2009, 18, 4442-4456.	1.4	99
90	The Novel SMAC Mimetic Birinapant Exhibits Potent Activity against Human Melanoma Cells. Clinical Cancer Research, 2013, 19, 1784-1794.	3.2	98

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91	Discovery and fine-mapping of adiposity loci using high density imputation of genome-wide association studies in individuals of African ancestry: African Ancestry Anthropometry Genetics Consortium. PLoS Genetics, 2017, 13, e1006719.	1.5	98
92	Refined histopathological predictors of BRCA1 and BRCA2mutation status: a large-scale analysis of breast cancer characteristics from the BCAC, CIMBA, and ENIGMA consortia. Breast Cancer Research, 2014, 16, 3419.	2.2	97
93	Application of a BRAF Pyrosequencing Assay for Mutation Detection and Copy Number Analysis in Malignant Melanoma. Journal of Molecular Diagnostics, 2007, 9, 464-471.	1.2	95
94	Identification of four novel susceptibility loci for oestrogen receptor negative breast cancer. Nature Communications, 2016, 7, 11375.	5.8	93
95	Interplay between BRCA1 and RHAMM Regulates Epithelial Apicobasal Polarization and May Influence Risk of Breast Cancer. PLoS Biology, 2011, 9, e1001199.	2.6	91
96	Cancer Screening Recommendations and Clinical Management of Inherited Gastrointestinal Cancer Syndromes in Childhood. Clinical Cancer Research, 2017, 23, e107-e114.	3.2	91
97	Identification of a Novel Subgroup of Melanomas with KIT/Cyclin-Dependent Kinase-4 Overexpression. Cancer Research, 2008, 68, 5743-5752.	0.4	90
98	Frequent genetic abnormalities of the PI3K/AKT pathway in primary ovarian cancer predict patient outcome. Genes Chromosomes and Cancer, 2011, 50, 606-618.	1.5	90
99	Genome-wide association and transcriptome studies identify target genes and risk loci for breast cancer. Nature Communications, 2019, 10, 1741.	5.8	90
100	Male breast cancer in BRCA1 and BRCA2 mutation carriers: pathology data from the Consortium of Investigators of Modifiers of BRCA1/2. Breast Cancer Research, 2016, 18, 15.	2.2	88
101	Shared heritability and functional enrichment across six solid cancers. Nature Communications, 2019, 10, 431.	5.8	88
102	Common Genetic Variants and Modification of Penetrance of BRCA2-Associated Breast Cancer. PLoS Genetics, 2010, 6, e1001183.	1.5	85
103	Arginase 2 Suppresses Renal Carcinoma Progression via Biosynthetic Cofactor Pyridoxal Phosphate Depletion and Increased Polyamine Toxicity. Cell Metabolism, 2018, 27, 1263-1280.e6.	7.2	85
104	Polygenic risk scores and breast and epithelial ovarian cancer risks for carriers of BRCA1 and BRCA2 pathogenic variants. Genetics in Medicine, 2020, 22, 1653-1666.	1.1	82
105	Restricted Expression of miR-30c-2-3p and miR-30a-3p in Clear Cell Renal Cell Carcinomas Enhances HIF2Î± Activity. Cancer Discovery, 2014, 4, 53-60.	7.7	79
106	Development of a tiered and binned genetic counseling model for informed consent in the era of multiplex testing for cancer susceptibility. Genetics in Medicine, 2015, 17, 485-492.	1.1	79
107	Genetic Subgrouping of Melanoma Reveals New Opportunities for Targeted Therapy: Figure 1.. Cancer Research, 2009, 69, 3241-3244.	0.4	78
108	Functional mechanisms underlying pleiotropic risk alleles at the 19p13.1 breastâ€“ovarian cancer susceptibility locus. Nature Communications, 2016, 7, 12675.	5.8	78

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109	BRCA2 Polymorphic Stop Codon K3326X and the Risk of Breast, Prostate, and Ovarian Cancers. Journal of the National Cancer Institute, 2016, 108, djv315.	3.0	77
110	Comprehensive characterization of the DNA amplification at 13q34 in human breast cancer reveals TFDP1 and CUL4A as likely candidate target genes. Breast Cancer Research, 2009, 11, R86.	2.2	75
111	Assessment of polygenic architecture and risk prediction based on common variants across fourteen cancers. Nature Communications, 2020, 11, 3353.	5.8	75
112	Immune Activation and a 9-Year Ongoing Complete Remission Following CD40 Antibody Therapy and Metastasectomy in a Patient with Metastatic Melanoma. Cancer Immunology Research, 2014, 2, 1051-1058.	1.6	74
113	Evaluation of linkage of breast cancer to the putative BRCA3 locus on chromosome 13q21 in 128 multiple case families from the Breast Cancer Linkage Consortium. Proceedings of the National Academy of Sciences of the United States of America, 2002, 99, 827-831.	3.3	73
114	Genetic and Genomic Characterization of 462 Melanoma Patient-Derived Xenografts, Tumor Biopsies, and Cell Lines. Cell Reports, 2017, 21, 1936-1952.	2.9	72
115	Common breast cancer susceptibility alleles are associated with tumour subtypes in BRCA1 and BRCA2 mutation carriers: results from the Consortium of Investigators of Modifiers of BRCA1/2. Breast Cancer Research, 2011, 13, R110.	2.2	71
116	Adjusting the estimated proportion of breast cancer cases associated with BRCA1 and BRCA2 mutations: Public health implications. Genetics in Medicine, 2005, 7, 28-33.	1.1	70
117	A genome-wide association study of breast cancer in women of African ancestry. Human Genetics, 2013, 132, 39-48.	1.8	70
118	Tumor Immunity and Survival as a Function of Alternative Neopeptides in Human Cancer. Cancer Immunology Research, 2018, 6, 276-287.	1.6	69
119	Common alleles at 6q25.1 and 1p11.2 are associated with breast cancer risk for BRCA1 and BRCA2 mutation carriers. Human Molecular Genetics, 2011, 20, 3304-3321.	1.4	68
120	BRCA1 and BRCA2 Mutation Frequency in Women Evaluated in a Breast Cancer Risk Evaluation Clinic. Journal of Clinical Oncology, 2002, 20, 994-999.	0.8	67
121	Evolution of delayed resistance to immunotherapy in a melanoma responder. Nature Medicine, 2021, 27, 985-992.	15.2	67
122	The APC I1307K allele and breast cancer risk. Nature Genetics, 1998, 20, 13-14.	9.4	65
123	Cancer Cell Lines as Genetic Models of Their Parent Histology: Analyses Based on Array Comparative Genomic Hybridization. Cancer Research, 2007, 67, 3594-3600.	0.4	65
124	Evaluation of 19 susceptibility loci of breast cancer in women of African ancestry. Carcinogenesis, 2012, 33, 835-840.	1.3	64
125	A Comparison of DNA Copy Number Profiling Platforms. Cancer Research, 2007, 67, 10173-10180.	0.4	62
126	CODEX2: full-spectrum copy number variation detection by high-throughput DNA sequencing. Genome Biology, 2018, 19, 202.	3.8	62

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127	Common variants associated with breast cancer in genome-wide association studies are modifiers of breast cancer risk in BRCA1 and BRCA2 mutation carriers. Human Molecular Genetics, 2010, 19, 2886-2897.	1.4	60
128	Genomic Signatures Predict the Immunogenicity of BRCA-Deficient Breast Cancer. Clinical Cancer Research, 2019, 25, 4363-4374.	3.2	60
129	Testicular germ cell tumor susceptibility associated with the UCK2 locus on chromosome 1q23. Human Molecular Genetics, 2013, 22, 2748-2753.	1.4	59
130	Uncommon Filaggrin Variants Are Associated with Persistent Atopic Dermatitis in African Americans. Journal of Investigative Dermatology, 2018, 138, 1501-1506.	0.3	59
131	Points to consider for reporting of germline variation in patients undergoing tumor testing: a statement of the American College of Medical Genetics and Genomics (ACMG). Genetics in Medicine, 2020, 22, 1142-1148.	1.1	59
132	Risk of metachronous breast cancer after BRCA mutationâ€“associated ovarian cancer. Cancer, 2013, 119, 1344-1348.	2.0	58
133	The Anti-Melanoma Activity of Dinaciclib, a Cyclin-Dependent Kinase Inhibitor, Is Dependent on p53 Signaling. PLoS ONE, 2013, 8, e59588.	1.1	58
134	Integrative Genomic Analyses of Sporadic Clear Cell Renal Cell Carcinoma Define Disease Subtypes and Potential New Therapeutic Targets. Cancer Research, 2012, 72, 112-121.	0.4	57
135	Associations of common breast cancer susceptibility alleles with risk of breast cancer subtypes in BRCA1 and BRCA2 mutation carriers. Breast Cancer Research, 2014, 16, 3416.	2.2	57
136	Patient feedback and early outcome data with a novel tiered-binned model for multiplex breast cancer susceptibility testing. Genetics in Medicine, 2016, 18, 25-33.	1.1	56
137	Multiple Endocrine Neoplasia and Hyperparathyroid-Jaw Tumor Syndromes: Clinical Features, Genetics, and Surveillance Recommendations in Childhood. Clinical Cancer Research, 2017, 23, e123-e132.	3.2	55
138	A patient-derived-xenograft platform to study BRCA-deficient ovarian cancers. JCI Insight, 2017, 2, e89760.	2.3	55
139	Common breast cancer risk variants in the post-COGS era: a comprehensive review. Breast Cancer Research, 2013, 15, 212.	2.2	52
140	A Multicenter Study of Cancer Incidence in CHEK2 1100delC Mutation Carriers: Table 1.. Cancer Epidemiology Biomarkers and Prevention, 2006, 15, 2542-2545.	1.1	51
141	Contribution of Germline Predisposition Gene Mutations to Breast Cancer Risk in African American Women. Journal of the National Cancer Institute, 2020, 112, 1213-1221.	3.0	51
142	Identification of intragenic deletions and duplication in the FLCN gene in Birtâ€Hoggâ€DubÃ© syndrome. Genes Chromosomes and Cancer, 2011, 50, 466-477.	1.5	50
143	Immunotherapy at Large: The road to personalized cancer vaccines. Nature Medicine, 2013, 19, 1098-1100.	15.2	50
144	Genome-wide association studies in women of African ancestry identified 3q26.21 as a novel susceptibility locus for oestrogen receptor negative breast cancer. Human Molecular Genetics, 2016, 25, ddw305.	1.4	50

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145	A practical guide for evaluating gonadal germ cell tumor predisposition in differences of sex development. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2017, 175, 304-314.	0.7	50
146	Modification of BRCA1-Associated Breast and Ovarian Cancer Risk by BRCA1-Interacting Genes. Cancer Research, 2011, 71, 5792-5805.	0.4	49
147	Characterization of the Cancer Spectrum in Men With GermlineBRCA1andBRCA2Pathogenic Variants. JAMA Oncology, 2020, 6, 1218.	3.4	48
148	The Role of KRAS rs61764370 in Invasive Epithelial Ovarian Cancer: Implications for Clinical Testing. Clinical Cancer Research, 2011, 17, 3742-3750.	3.2	47
149	Common Variants at the 19p13.1 and ZNF365 Loci Are Associated with ER Subtypes of Breast Cancer and Ovarian Cancer Risk in BRCA1 and BRCA2 Mutation Carriers. Cancer Epidemiology Biomarkers and Prevention, 2012, 21, 645-657.	1.1	47
150	Risk of Breast Cancer Among Carriers of Pathogenic Variants in Breast Cancer Predisposition Genes Varies by Polygenic Risk Score. Journal of Clinical Oncology, 2021, 39, 2564-2573.	0.8	47
151	Genetic variation in insulin-like growth factor signaling genes and breast cancer risk among BRCA1 and BRCA2 carriers. Breast Cancer Research, 2009, 11, R76.	2.2	44
152	A functionally significant SNP in TP53 and breast cancer risk in African-American women. Npj Breast Cancer, 2017, 3, 5.	2.3	44
153	Successful Use of Alternate Waste Nitrogen Agents and Hemodialysis in a Patient With Hyperammonemic Coma After Heart-Lung Transplantation. Archives of Neurology, 1999, 56, 481.	4.9	43
154	Association of Inherited Pathogenic Variants in Checkpoint Kinase 2 (CHEK2) With Susceptibility to Testicular Germ Cell Tumors. JAMA Oncology, 2019, 5, 514.	3.4	43
155	The International Testicular Cancer Linkage Consortium: A clinicopathologic descriptive analysis of 461 familial malignant testicular germ cell tumor kindred. Urologic Oncology: Seminars and Original Investigations, 2010, 28, 492-499.	0.8	42
156	Inheritance of deleterious mutations at both BRCA1 and BRCA2 in an international sample of 32,295 women. Breast Cancer Research, 2016, 18, 112.	2.2	42
157	Genomic Biomarkers for Breast Cancer Risk. Advances in Experimental Medicine and Biology, 2016, 882, 1-32.	0.8	42
158	Genetic variants demonstrating flip-flop phenomenon and breast cancer risk prediction among women of African ancestry. Breast Cancer Research and Treatment, 2018, 168, 703-712.	1.1	42
159	A Recurrent ERCC3 Truncating Mutation Confers Moderate Risk for Breast Cancer. Cancer Discovery, 2016, 6, 1267-1275.	7.7	41
160	Evaluating Polygenic Risk Scores for Breast Cancer in Women of African Ancestry. Journal of the National Cancer Institute, 2021, 113, 1168-1176.	3.0	41
161	Genetic Variation at 9p22.2 and Ovarian Cancer Risk for BRCA1 and BRCA2 Mutation Carriers. Journal of the National Cancer Institute, 2011, 103, 105-116.	3.0	40
162	The Molecular Biology of Renal Cell Carcinoma. Seminars in Oncology, 2013, 40, 421-428.	0.8	40

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163	Functional characterization of a multi-cancer risk locus on chr5p15.33 reveals regulation of TERT by ZNF148. Nature Communications, 2017, 8, 15034.	5.8	40
164	Predicting Metastatic Potential in Pheochromocytoma and Paraganglioma: A Comparison of PASS and GAPP Scoring Systems. Journal of Clinical Endocrinology and Metabolism, 2020, 105, e4661-e4670.	1.8	40
165	Association of Filaggrin Loss-of-Function Variants With Race in Children With Atopic Dermatitis. JAMA Dermatology, 2019, 155, 1269.	2.0	39
166	Association of Genomic Domains in BRCA1 and BRCA2 with Prostate Cancer Risk and Aggressiveness. Cancer Research, 2020, 80, 624-638.	0.4	39
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